SERPINA7 gene

serpin family A member 7

Normal Function

The *SERPINA7* gene (also known as *TBG*) provides instructions for making a protein called thyroxine-binding globulin. In the bloodstream, this protein carries hormones made or used by the thyroid gland, which is a butterfly-shaped tissue in the lower neck. Thyroid hormones play an important role in regulating growth, brain development, and the rate of chemical reactions in the body (metabolism). Most of the time, thyroid hormones circulate in the bloodstream attached to thyroxine-binding globulin and similar proteins.

Health Conditions Related to Genetic Changes

inherited thyroxine-binding globulin deficiency

More than 25 mutations in the *SERPINA7* gene have been identified in people with inherited thyroxine-binding globulin deficiency. Some mutations lead to a shortened, nonfunctional version of thyroxine-binding globulin. These genetic changes result in a total loss of the protein, which causes the complete form of inherited thyroxine-binding globulin deficiency (TBG-CD). Other mutations change single protein building blocks (amino acids) in thyroxine-binding globulin. These mutations alter the structure or processing of the protein, leading to the partial form of the disorder (TBG-PD).

When there is a shortage of thyroxine-binding globulin, the amount of circulating thyroid hormones is reduced. These changes do not cause any problems with thyroid function. Although inherited thyroxine-binding globulin deficiency does not cause any health problems, it can be mistaken for more serious thyroid disorders (such as hypothyroidism). Therefore, it is important to diagnose inherited thyroxine-binding globulin deficiency to avoid unnecessary treatments.

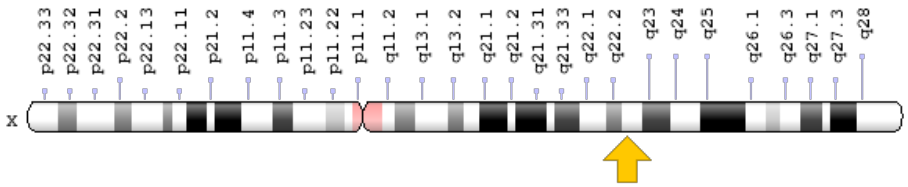
other disorders

Other changes involving the *SERPINA7* gene cause a condition called thyroxine-binding globulin excess (TBG-E). People with this condition have unusually high levels of thyroxine-binding globulin, often two to four times greater than normal. This excess is caused by the presence of one or more extra copies of the *SERPINA7* gene in each cell. Like thyroxine-binding globulin deficiency, thyroxine-binding globulin excess does not cause any problems with thyroid function.

Chromosomal Location

Cytogenetic Location: Xq22.3, which is the long (q) arm of the X chromosome at position 22.3

Molecular Location: base pairs 106,032,439 to 106,038,727 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alpha-1 antiproteinase, antitrypsin
- serine (or cysteine) proteinase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 7
- serine (or cysteine) proteinase inhibitor, clade A, member 7
- serpin peptidase inhibitor, clade A (alpha-1 antiproteinase, antitrypsin), member 7
- TBG
- THBG_HUMAN
- thyroxin-binding globulin
- thyroxine-binding globulin

Additional Information & Resources

Educational Resources

- Endocrinology (2001): Transport and metabolism of thyroid hormones https://www.ncbi.nlm.nih.gov/books/NBK28/#A393

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28SERPINA7%5BTIAB%5D%29+OR+%28%28TBG%5BTI%5D%29+OR+%28thyroxin-binding+globulin%5BTI%5D%29+OR+%28thyroxine-binding+globulin%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3240+days%22%5Bdp%5D

OMIM

- THYROXINE-BINDING GLOBULIN OF SERUM
  http://omim.org/entry/314200

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SERPINA7%5Bgene%5D
- HGNC Gene Family: Serpin peptidase inhibitors
  http://www.genenames.org/cgi-bin/genefamilies/set/739
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11583
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6906
- UniProt
  http://www.uniprot.org/uniprot/P05543

Sources for This Summary

- Domingues R, Font P, Sobrinho L, Bugalho MJ. A novel variant in Serpina7 gene in a family with thyroxine-binding globulin deficiency. Endocrine. 2009 Aug;36(1):83-6. doi: 10.1007/s12020-009-9202-2. Epub 2009 May 5.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19415532
- Hayashi Y, Mori Y, Janssen OE, Sunthornthepvarakul T, Weiss RE, Takeda K, Weinberg M, Seo H, Bell GI, Refetoff S. Human thyroxine-binding globulin gene: complete sequence and transcriptional regulation. Mol Endocrinol. 1993 Aug;7(8):1049-60.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8232304
- Mori Y, Miura Y, Takeuchi H, Igarashi Y, Sugiura J, Saito H, Oiso Y. Gene amplification as a cause of inherited thyroxine-binding globulin excess in two Japanese families. J Clin Endocrinol Metab. 1995 Dec;80(12):3758-62.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8530630
- Schussler GC. The thyroxine-binding proteins. Thyroid. 2000 Feb;10(2):141-9. Review. Erratum in: Thyroid 2000 Apr;10(4):372.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10718550

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/SERPINA7

Reviewed: September 2009
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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